RPS6KA3 gene

ribosomal protein S6 kinase A3

Normal Function

The *RPS6KA3* gene provides instructions for making a protein that is part of a family called ribosomal S6 kinases (RSKs). These proteins help regulate the activity of certain genes and are involved in signaling within cells. RSK proteins are thought to play a role in several important cellular processes including cell growth and division (proliferation), cell specialization (differentiation), and the self-destruction of cells (apoptosis).

The protein made by the *RPS6KA3* gene appears to play an important role in the brain. The protein is involved in cell signaling pathways that are required for learning, the formation of long-term memories, and the survival of nerve cells.

Health Conditions Related to Genetic Changes

Coffin-Lowry syndrome

More than 125 mutations in the *RPS6KA3* gene have been identified in people with Coffin-Lowry syndrome, a condition associated with intellectual disability and skeletal abnormalities. All of these mutations severely reduce or eliminate the activity of the RPS6KA3 protein. Some mutations insert or delete genetic material in the gene or change how the gene's instructions are used to build the protein. Other mutations change single protein building blocks (amino acids) in the RPS6KA3 protein.

Researchers do not fully understand how mutations in the *RPS6KA3* gene lead to the signs and symptoms of Coffin-Lowry syndrome. A functional RPS6KA3 protein appears to be important for learning and memory, but its role in the skeleton is unknown.

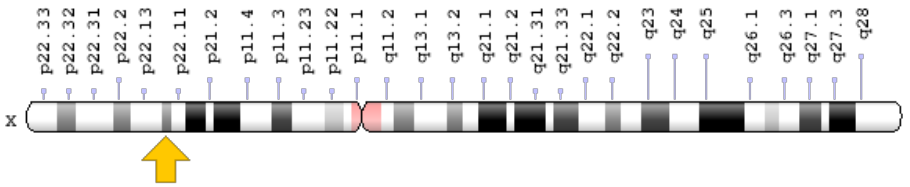
other disorders

RPS6KA3 mutations have been identified in some people who have intellectual disability, but do not have most of the other characteristic features of Coffin-Lowry syndrome. Because the *RPS6KA3* gene is on the X chromosome, this condition is known as X-linked intellectual disability. Researchers believe that *RPS6KA3* mutations are a rare cause of intellectual disability because only a few affected families have mutations in this gene.

Chromosomal Location

Cytogenetic Location: Xp22.12, which is the short (p) arm of the X chromosome at position 22.12

Molecular Location: base pairs 20,149,911 to 20,267,514 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- HU-2
- HU-3
- Insulin-stimulated protein kinase 1
- ISPK-1
- KS6A3_HUMAN
- MAP kinase-activated protein kinase 1b
- MAPKAPK1B
- MRX19
- p90(rsk)
- ribosomal protein S6 kinase, 90kDa, polypeptide 3
- Ribosomal S6 kinase 2
- RSK-2
- RSK2

Additional Information & Resources

GeneReviews

- Coffin-Lowry Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1346

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28RPS6KA3%5BTIAB%5D%29+OR+%28RSK2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- RIBOSOMAL PROTEIN S6 KINASE, 90-KD, 3
  http://omim.org/entry/300075

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_RPS6KA3.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=RPS6KA3%5Bgene%5D
- HGNC Gene Family: Mitogen-activated protein kinase-activated protein kinases
  http://www.genenames.org/cgi-bin/genefamilies/set/1156
- HGNC Gene Family: X-linked mental retardation
  http://www.genenames.org/cgi-bin/genefamilies/set/103
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10432
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6197
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